4.

Mr. Marshall
with Thos Barlow's
kind regards

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Limb-bones, skull, and brain of a case of so-called fœtal rickets (? fœtal cretinism).

By Thomas Barlow, M.D.

[With Plate XXXIII, fig. 1.]

The body from which these specimens were taken was sent to me by my friend Dr. Colegate, of Eastbourne. It was full term, and born of a healthy mother, who had previously given birth to five healthy children. It was extremely blue at birth, and only breathed for a few seconds.

Dr. Colegate, noticing the short stunted limbs with transverse folds, and the general accumulation of subcutaneous fat, suspected that it would present the same osseous characters as those found in the case shown by me at the rickets debate two years ago ("So-called Fœtal Rickets," 'Path. Trans.,' vol. xxxii, p. 364).

The resemblances in many points are so remarkable that portions of the respective skeletons are almost indistinguishable.

With regard to both cases, it may be briefly stated that the essential feature in the long bones is a defective row formation in the primordial cartilage cells, the earthy salts being consequently deposited right across, but not in vertical lines. This fundamental defect has to do with the stunting in length growth of the limbs, the principal *rôle* in ossification being performed by the periosteum. The bones formed in membrane are, on the other hand, well developed and proportioned; thus, taking the shoulder-girdle, the clavicle, which is a membrane-formed bone, is well developed. The scapula shows a marked overlapping of the epiphysial cartilage by bony sheaths. The humerus shows relatively large epiphyses and a short, stout shaft. That the epiphyses are only relatively large will be seen by comparison with the accompanying humerus from a healthy full-term fœtus.

There is no proliferation of cartilage forming digitate processes along the line of ossification, such as one sees in true rickets. Along the upper line is a fibrous lamina, which has grown in from the periosteum.

There is a premature ossifying centre in the upper epiphysis,

quite eccentrically situated, viz. just above the fibrous lamina referred to, and probably arising in connection with this fibrous tissue invasion.

The shaft is short and stout, and presents a marked concavity forwards, with projection backwards at the lower end.

The lower line of ossification is regular, but unnaturally convex. There is no fibrous invasion here, and no nucleus in the lower epiphysis.

The radius presents the same remarkable sigmoid curve, an exaggeration of the normal one, which was found in the former case (*vide* fig. 1, p. 365, 'Path. Trans.,' vol. xxxii).

The hand is short and stunted, and there is considerable accumulation of subcutaneous fat.

The pelvis and lower limbs show similar characters to those of the first case, and in like manner the ribs present beads at the junction with the cartilages, which are formed, not by proliferated cartilage gradually becoming ossified, as in true rickets, but by an investing sheath of bone round the end of the costal cartilage, derived, no doubt, from the periosteum of the rib.

There are no ossifying centres in the sternum. The vertebræ are normal.

The skull presents the same remarkable shortening in the basis cranii which is formed in cartilage. The basi-occipital and basi sphenoid are prematurely ankylosed, and so are the basi-sphenoid and præ-sphenoid. The clivus is very steep, the foramen magnum funnel shaped, the cartilage-formed portion of the occiput extremely stunted, whilst the portion formed in membrane has undergone extra development. In every respect this skull is almost a replica of the former specimen.

I will now refer to the brain, which presents some remarkable features at the base, in relation to the malformation of the skull. The crura cerebri, pons, and medulla are more nearly vertical in their direction than obtains in the normal brain, and the pons is laterally compressed. These are all in relation to the steep clivus and the deep funnel-shaped foramen magnum. The cerebellum is more covered by the cerebrum than in a healthy fœtal brain. Dividing the antero-posterior axis into four equal parts the cerebellum occupies the third quarter of it. This is related to the smallness of the occiput below the internal protuberance, which has had the effect of eliminating the cerebellar fossæ. The cerebellum

has, therefore, been pushed forwards, and its growth has been much more in an upward direction than is normal. Thus, when the brain is looked at with its base upwards, the cerebellum seems to be completely nested in the cerebrum. It is also to be noted that the right hemisphere of the cerebellum is a little less than the left.

There is nothing remarkable about the arrangement of the convolutions of the cerebral hemispheres on the convexity, but on the inferior surface it is very remarkable indeed, and, so far as I know, quite unique. The exceptional features concern the lower part of the temporo-sphenoidal lobes. On the left side, the inferior temporo-sphenoidal convolution and the temporo-occipital convolution are divided into two parts by a very deep fissure. The commencement of this fissure may be indicated as to locality by its proximity to what may be called the most external portion of the left hemisphere of the cerebellum; that is to say, a point at the junction of the anterior third with the posterior two thirds of its circumference. This fissure passes from proximity to the above point in a slightly curved direction forwards and upwards as far as the middle temporo-sphenoidal convolution to within three quarters of an inch of the posterior limb of the fissure of Sylvius, to which it lies almost at right angles. Besides splitting the inferior temporo-sphenoidal and the temporo-occipital convolutions it also divides the uncinate convolution. At the bottom of the fissure there are situated a number of small botryoidal eminences, due to subdivision of the uncinate convolution. On the right side there is a similar fissure. Also on the right side, the surface of the portion of the temporo-sphenoidal lobe which is anterior to the deep fissure is marked by small fissures, having a general direction forwards and outwards parallel to the deep fissure. One of these, considerably deeper than the others, begins at the margin, about one third of an inch in front of the large fissure, and approximately divides this portion of the lobe into two.

I have failed to find anything in the normal fœtal brain with which these fissures correspond. It seems probable that they are related to the upward thrust of the cerebellum.

There is nothing abnormal about the corpus callosum, fornix, or the great ganglia.

With respect to the viscera, I need only refer to the heart, which presents some remarkable malformations.

In the right auricle there is a large patent foramen ovale. There

is a large right auriculo-ventricular orifice, with a well-formed tricuspid valve. The pulmonary orifice is guarded by two equal valves, which are united, so that they make a funnel-shaped opening. The pulmonary artery is a larger vessel than the aorta.

There is a hole through the "undefended spot" at the top of the septum, large enough to admit of a pea, and opening into the left ventricle. Just above this opening, and communicating thus with both right and left ventricles, arises the aorta. As far as the specimen shows, the aorta appears to have been furnished with a pair of valves, similar to the pulmonary. A thin cribriform membrane is attached above by fine fibrous processes to the edge and under surface of the posterior aortic valve and to part of the septal segment of the tricuspid. This membrane arises from a small musculus papillaris, in common with that from which the septal segment of the tricuspid arises. It would appear to be the aborted representative of the mitral valve. The left ventricle is much smaller than the right; it has no mitral orifice, but only the orifice through the undefended spot. The left auricle receives the pulmonary veins, but has no orifice except the patent foramen ovale.

The thyroid gland was natural to naked-eye inspection.

There were no separate fat masses in the neck, but there was a general accumulation of fat in the subcutaneous areolar tissue, to which I have already referred.

Remarks.—In my former paper I have shown that cases of this kind, although sometimes described under the category of fœtal rickets, differ essentially from true rickets, and that they ought to be relegated to a class of malformations depending on a very early vice of development.

As a question of nomenclature, it may be asked whether it is proper to group these cases with cretins.

In favour of this classification are the general features, viz., the stunted limbs compared with the relatively large head and belly, the depressed nose root, and the heaping up of subcutaneous fat.

The picture and description given by Virchow of a newborn cretin, the offspring of a cretin mother,[1] in many respects strikingly corresponds with the two cases that I have shown—most notably

[1] P. 976, 'Gesammelte Abhandlungen zur Wissenschaftlichen Medicin,' 1856.

in regard to the synostosis of the basis cranii, which Virchow was the first to discover.[1]

Furthermore, the "calf cretins" described by Eberth[2] have many allied characters, including not only the cranial synostosis, but the special histological conditions of the growing ends of the long bones to which I have referred.

I must, however, point out that these cases differ in many respects from the sporadic cretins described in England by Mr. Curling, and subsequently by Dr. Hilton Fagge. In the latter cases, of which I have seen a goodly number, although there was stunted growth, there was no such remarkable arrest in the length growth of the limbs, as is manifest in Virchow's specimen and the one now exhibited. Further, the localised fat masses first described by Mr. Curling were not present in my two specimens, and are not referred to by Virchow. Also with regard to the thyroid, Virchow describes it as being enlarged in his case, and in my specimens it was certainly present. Dr. Fagge laid great stress on the absence of the isthmus of the thyroid in his sporadic cases, and was inclined at first to make this a distinguishing mark between sporadic and endemic cretins. It is to be remembered, however, that one of his cases in which during life the isthmus of the thyroid was believed to be absent was found on *post-mortem* examination to have a goître.[3]

With respect to the basis cranii, the age of the patient examined *post-mortem* by Dr. Fagge rendered it impossible to say whether there had been premature synostosis or not, but the clivus, instead of being steeper, was more horizontal than normal. In a sporadic cretin, examined *post-mortem* by Dr. Abercrombie and myself, the synchondrosis between the basi-occipital and basi-sphenoid was still present.

To sum up, I conceive that in calling my two specimens cretins I am following the lead of Virchow and Eberth.

Perhaps these cases may be regarded as belonging to a *very pronounced fœtal type of cretinism.* It is, I think, doubtful whether any human cretin of this kind has survived birth. It is much to be desired that cases of the type described by Dr. Fagge should

[1] See also account of a specimen in the Berlin collection which had been labelled Congenital Rickets, 'Virchow's Archiv,' 1858, p. 353.

[2] Eberth 'Die Fötale Rachitis und ihre Beziehungen zu dem Cretinismus,' 1878.

[3] 'Path. Trans.,' 1874, p. 268.

be examined *post-mortem*, especially with regard to the ossification of the long bones. It seems possible that there may be varying degrees of the same faulty development along the growing edge corresponding with more or less dwarfing of limbs and disproportion in their length to that of the trunk. *Feb. 5th*, 1884.

DESCRIPTION OF PLATE XXXIII.

Fig. 1.—To illustrate Dr. Barlow's case of Brain from a Cretinous Fœtus.

It shows the almost vertical direction of the medulla, the cerebellum pushed upwards into the cerebrum, and remarkable abnormal fissures of the temporo-spenoidal lobes.